

# Genodermatoses: Basic and Practical Aspects

Residents Course | 12-13 January 2023 | Innsbruck, Austria

## **Faculty**

**Course Chairs:** Matthias Schmuth, Johann Bauer

**Speakers:** Johannes Zschocke, Judith Fischer, Robert Gruber

**Tutors:** Verena Moosbrugger-Martinz, Daniela Ortner-Tobider, Thomas Trafoier

## **Course Description**

The genodermatoses course is designed to introduce trainees to the increasingly important role of genetics and genomics in dermatology. Teaching is provided by a diverse faculty consisting of dermatologists, geneticists and pediatricians to reflect the interdisciplinary approach necessary to best care for patients with genodermatosis.

The course includes both lectures and a laboratory tour for all participants. In past courses, participants commented that the laboratory part was an eye-opening experience, which greatly helped them to truly grasp the nature of the diagnostic process involved in identifying gene mutations in genodermatoses. There will also be time for case discussions and social interaction, i.e., networking among the residents and specialists from many different countries.

Genodermatoses are rare diseases (orphan diseases) with less than 1 in 2.000 people affected among the general population. Genodermatoses can be debilitating, severely affect the quality of life, and they may affect other organ systems. Without treatment, they often result in reduced life expectancy. Although individual entities are rare, the sum of all patients with genodermatoses represents a considerable number of patients who need specialty care. Thus, it is of major importance to teach the physicians of our specialty about these conditions requiring specific care due to the difficulty in establishing a diagnosis. The course also will address measures to prevent complications and to how set up treatment and patient management programs. In the field of rare diseases, it is of tremendous importance to share and transmit the expertise, for example through EADV training courses.

## **Learning Objectives**

1. Understand genodermatoses within spectrum of rare diseases in humans
2. Learn clinical differential diagnosis of genodermatoses
3. Determine molecular basis and therapy of genodermatoses

## **Programme**

### **Thursday, 12 January 2023**

- 08:30-09:00 Arrival and welcome
- 09:00-10:00 Genetics 101: The Basics – Johannes Zschocke
- 10:00-10:30 **Coffee break**
- 10:30-11:30 Non-syndromic Ichthyoses – Judith Fischer
- 11:30-12:00 Update neurofibromatosis type 1 – Robert Gruber
- 12:00-13:00 **Lunch break**
- 13:00-14:00 Epidermolysis bullosa: Molecular Basis – Johann Bauer
- 14:00-15:00 Genetic diagnostics: Interactive cases – Verena Moosbrugger-Martinz
- 15:00-15:30 **Coffee break**
- 15:30-16:00 Syndromic Ichthyosis – Judith Fischer
- 16:00-16:30 Epidermolysis bullosa: Therapy – Johann Bauer
- 16:30-17:30 Connective Tissue Diseases – Johannes Zschocke
- 19:30-22:00 **Networking dinner with faculty and participants**

### **Friday, 13 January 2023**

- 08:00-09:00 Practical part: Visit of testing facilities – Johannes Zschocke, Robert Gruber, Daniela Ortner-Tobider, Verena Moosbrugger-Martinz, Thomas Trafoier
- 09:00-10:00 Quiz, case reports – Participants, Daniela Ortner-Tobider, Verena Moosbrugger-Martinz, Thomas Trafoier
- 10:00-10:30 **Coffee break**
- 10:30-11:00 Palmoplantar Keratoderma – Matthias Schmuth
- 11:00-12:00 Differential diagnosis of syndromes with facial skin lesions – Matthias Schmuth
- 12:00-13:00 **Lunch break**
- 13:00-14:00 Genodermatoses with dental involvement – Robert Gruber, Johannes Zschocke
- 14:00-14:30 Cornification disorders: Therapy – Matthias Schmuth
- 14:30-15:00 Porphyria – Matthias Schmuth
- 15:00-15:30 **Coffee break**
- 15:30-17:30 Quiz, participants´ case reports and discussion – Daniela Ortner-Tobider, Verena Moosbrugger-Martinz, Thomas Trafoier